

WHAT IS CLAIMED IS:

1. A method for detecting the presence or diagnosing the risk of prostate cancer in a patient, comprising detecting in a biological sample obtained from the patient a level or functional activity of an aberrant expression product of a gene selected from *PSA* or *KLK2*, which correlates with the presence or risk of prostate cancer.
2. The method of claim 1, wherein the aberrant *PSA* expression product is selected from an aberrant *PSA* transcript or polypeptide product thereof, wherein the transcript comprises a 145 nt insertion from intron 3 of the wild-type *PSA* gene.
3. The method of claim 1, wherein the aberrant *PSA* expression product is selected from *PSA* RP2 transcript 1, *PSA* RP2 transcript 2, or a polypeptide product of these.
4. The method of claim 1, wherein the aberrant *PSA* expression product is a transcript comprising the sequence set forth in SEQ ID NO: 7.
5. The method of claim 1, wherein the aberrant *PSA* expression product is a transcript comprising the sequence set forth in SEQ ID NO: 1.
6. The method of claim 1, wherein the aberrant *PSA* expression product is a transcript comprising the sequence set forth in SEQ ID NO: 3.
7. The method of claim 1, wherein the aberrant *PSA* expression product is a transcript comprising the sequence set forth in SEQ ID NO: 5.
8. The method of claim 1, wherein the aberrant *PSA* expression product is a polypeptide comprising a truncated prepro-PSA variant.
9. The method of claim 1, wherein the aberrant *PSA* expression product is a polypeptide having reduced serine protease activity relative to wild-type PSA.
10. The method of claim 1, wherein the aberrant *PSA* expression product is a polypeptide that is devoid of serine protease activity.
11. The method of claim 1, wherein the aberrant *PSA* expression product is a polypeptide comprising the sequence set forth in SEQ ID NO: 2.
12. The method of claim 1, wherein the aberrant *PSA* expression product is a polypeptide comprising the sequence set forth in SEQ ID NO: 4.
13. The method of claim 1, wherein the aberrant *PSA* expression product is a polypeptide comprising the sequence set forth in SEQ ID NO: 6.

14. The method of claim 1, wherein the aberrant *KLK2* expression product is selected from an aberrant *KLK2* transcript or polypeptide product thereof, wherein the transcript comprises an intronic insertion and preferably a 37 nt insertion from intron 4 of the wild-type *KLK2* gene.

15. The method of claim 1, wherein the aberrant *KLK2* expression product is selected from *KLK2* 10A transcript or a polypeptide product thereof.

16. The method of claim 1, wherein the aberrant *KLK2* expression product is a transcript comprising the sequence set forth in SEQ ID NO: 9.

17. The method of claim 1, wherein the aberrant *KLK2* expression product is a transcript comprising the sequence set forth in SEQ ID NO: 11.

18. The method of claim 1, wherein the aberrant *KLK2* expression product is a transcript comprising the sequence set forth in SEQ ID NO: 13.

19. The method of claim 1, wherein the aberrant *KLK2* expression product is a polypeptide having reduced serine protease activity relative to wild-type K2.

20. The method of claim 1, wherein the aberrant *KLK2* expression product is a polypeptide that is devoid of serine protease activity.

21. The method of claim 1, wherein the aberrant *KLK2* expression product is a polypeptide comprising the sequence set forth in SEQ ID NO: 10.

22. The method of claim 1, wherein the aberrant *KLK2* expression product is a polypeptide comprising the sequence set forth in SEQ ID NO: 12.

23. The method of claim 1, wherein the aberrant *KLK2* expression product is a polypeptide comprising the sequence set forth in SEQ ID NO: 14.

24. The method of claim 1, wherein the correlation with the presence or risk of prostate cancer is made when the level or functional activity of the *PSA* or *KLK2* aberrant expression product in the biological sample is at least 10% higher than a reference level or functional activity of the expression product that correlates with the presence or risk of BPH.

25. The method of claim 1, further comprising quantifying an aberrant polypeptide product of the gene.

26. The method of claim 1, further comprising quantifying an aberrant polypeptide product of the gene, wherein the aberrant polypeptide product is quantified by:

- contacting the biological sample with an antigen-binding molecule that is immuno-interactive with the aberrant polypeptide product;
- measuring the concentration of a complex comprising the aberrant polypeptide product and the antigen binding molecule in the contacted sample; and

— relating the measured complex concentration to the concentration of the aberrant polypeptide product in the sample.

27. The method of claim 26, wherein the concentration of the aberrant polypeptide product in the biological sample is compared to a reference level of the aberrant polypeptide product which correlates with the presence or risk of BPH.

28. The method of claim 1, further comprising measuring the level of an aberrant transcript expressed from the gene in the biological sample.

29. The method of claim 28, wherein the level of the transcript in the biological sample is compared to a reference level of the transcript polypeptide which correlates with the presence or risk of BPH.

30. The method of claim 28, wherein the level of the transcript is measured using a probe that comprises a nucleotide sequence which corresponds or is complementary to at least a portion of the aberrant transcript.

31. The method of claim 28, wherein the level of an aberrant transcript is quantified using a nucleic acid amplification technique that quantifies the aberrant transcript in real-time.

32. The method of claim 1, comprising indirectly analysing the level of an aberrant polypeptide product of the gene.

33. The method of claim 1, comprising indirectly analysing the level of an aberrant polypeptide product of the gene by qualitatively or quantitatively determining in the biological sample the level of an antigen-binding molecule that is immuno-interactive with the aberrant polypeptide product.

34. The method of claim 1, comprising:

- contacting the biological sample with an antigen corresponding to at least a portion of the aberrant polypeptide product;
- measuring the concentration of a complex comprising the antigen and an antigen-binding molecule in the contacted sample; and
- relating the measured complex concentration to the concentration of antigen-binding molecule in the sample to thereby determine the amount or level of the aberrant polypeptide product in the sample.

35. The method of claim 1, wherein the prostate cancer is metastatic prostate cancer.

36. The method of claim 35, wherein the metastatic prostate cancer is associated with metastasis to a bone or lymph node of the patient.

37. The method of claim 1, wherein the expression product is present intracellularly.

38. The method of claim 1, wherein the expression product is present in soluble form.
39. The method of claim 38, wherein, the biological sample comprises a biological fluid selected from seminal fluid, whole blood, serum or lymphatic fluid.
40. The method of claim 1, wherein the prostate cancer is an organ-confined prostate cancer.
41. A method for detecting the presence or diagnosing the risk of prostate cancer in a patient, comprising detecting in a biological sample obtained from the patient a level or functional activity of an aberrant expression product of a gene selected from *PSA* or *KLK2*, which is higher than a reference level or functional activity of the expression product polypeptide which correlates with the presence or risk of benign prostatic hyperplasia (BPH).
42. A method for detecting the presence or diagnosing the risk of prostate cancer in a patient, comprising detecting in a biological sample obtained from the patient a level or functional activity of an aberrant expression product selected from *PSA* RP2 transcript 1, *PSA* RP2 transcript 2, *KLK2* 10A transcript or a polypeptide encoded thereby, which correlates with the presence or risk of prostate cancer.
43. A method for detecting the presence or diagnosing the risk of prostate cancer in a patient, comprising detecting in a biological sample obtained from the patient a level or functional activity of an aberrant expression product selected from *PSA* RP2 transcript 2, *KLK2* 10A transcript or a polypeptide encoded thereby, which correlates with the presence or risk of prostate cancer.
44. A method for detecting the presence or diagnosing the risk of prostate cancer in a patient, comprising detecting in a biological sample obtained from the patient a level or functional activity of a transcript selected from *PSA* RP2 transcript 1, *PSA* RP2 transcript 2 or *KLK2* 10A, which correlates with the presence or risk of prostate cancer.
45. A method for detecting the presence or diagnosing the risk of prostate cancer in a patient, comprising detecting in a biological sample obtained from the patient a level or functional activity of a transcript selected from *PSA* RP2 transcript 2 or *KLK2* 10A, which correlates with the presence or risk of prostate cancer.
46. A method for detecting the presence or diagnosing the risk of metastatic prostate cancer in a patient, comprising detecting in a biological sample obtained from the patient a level or functional activity of an aberrant expression product of a gene selected from *PSA* and *KLK2*, which correlates with the presence or risk of prostate cancer, wherein the biological sample comprises a fluid or tissue other than prostate tissue.
47. A method for detecting the presence or diagnosing the risk of organ-confined prostate cancer in a patient, comprising detecting in a biological sample obtained from the patient the

absence of a level or functional activity of an aberrant expression product of a gene selected from *PSA* and *KLK2*, which correlates with the presence or risk of prostate cancer, wherein the biological sample comprises a fluid or tissue other than prostate tissue.

48. Use of at least a portion of an aberrant expression product of a gene selected from *PSA* or *KLK2* in the manufacture of a kit for qualitatively or quantitatively determining a level or functional activity of the aberrant expression product, which correlates with the presence or risk of prostate cancer.

49. Use of a probe comprising a nucleotide sequence which corresponds or is complementary to at least a portion of an aberrant transcript of a gene selected from *PSA* or *KLK2* in the manufacture of a kit for qualitatively or quantitatively determining a level or functional activity of the aberrant transcript, which correlates with the presence or risk of prostate cancer.

50. Use of an antigen-binding molecule that is immuno-interactive specifically with an aberrant polypeptide product of a gene selected from *PSA* or *KLK2* in the manufacture of a kit for qualitatively or quantitatively determining a level or functional activity of the aberrant polypeptide product, which correlates with the presence or risk of prostate cancer.

51. Use of an agent in the manufacture of a medicament for treating and/or preventing prostate cancer, wherein the agent is selected from an antisense oligonucleotide, a ribozyme or an RNAi-mediating molecule that binds to, or otherwise interacts specifically with, an aberrant transcript of a gene selected from *PSA* or *KLK2*, whose level or functional activity correlates with the presence or risk of prostate cancer, wherein the agent is optionally formulated with a pharmaceutically acceptable carrier.

52. Use of an antigen-binding molecule that is immuno-interactive with an aberrant polypeptide product of a gene selected from *PSA* or *KLK2* whose level or functional activity correlates with the presence or risk of prostate cancer in the manufacture of a medicament for treating and/or preventing prostate cancer, wherein the agent is optionally formulated with a pharmaceutically acceptable carrier.

53. Use of an antigen-presenting cell in the manufacture of a medicament for modulating the level or functional activity of an aberrant polypeptide product of a gene selected from *PSA* or *KLK2*, or a variant or derivative of the polypeptide product, wherein the antigen-presenting cell expresses a processed form of the aberrant polypeptide product for presentation to, and modulation of, T cells.

54. A method for the treatment and/or prophylaxis of a prostate cancer, comprising administering to a patient in need thereof an effective amount of an agent selected from an antisense oligonucleotide, a ribozyme or an RNAi-mediating molecule that binds to, or otherwise

interacts specifically with, an aberrant transcript of a gene selected from *PSA* or *KLK2*, whose level or functional activity correlates with the presence or risk of prostate cancer, wherein the agent is optionally formulated with a pharmaceutically acceptable carrier.

55. A method for the treatment and/or prophylaxis of a prostate cancer, comprising administering to a patient in need thereof an effective amount of an antigen-binding molecule that is immuno-interactive with an aberrant polypeptide product of a gene selected from *PSA* or *KLK2* whose level or functional activity correlates with the presence or risk of prostate cancer, wherein the antigen-binding molecule is optionally formulated with a pharmaceutically acceptable carrier.

56. A method for the treatment and/or prophylaxis of a prostate cancer, comprising administering to a patient in need thereof an effective amount of an antigen-binding molecule that is immuno-interactive with an aberrant polypeptide product of a gene selected from *PSA* or *KLK2* whose level or functional activity correlates with the presence or risk of prostate cancer, wherein the antigen-binding molecule is optionally formulated with a pharmaceutically acceptable carrier.

57. A method for the treatment and/or prophylaxis of a prostate cancer, comprising administering to a patient in need thereof an effective amount of an antigen-presenting cell expressing a processed form of an aberrant polypeptide product of a gene selected from *PSA* or *KLK2* for presentation to, and modulation of, T cells, which level or functional activity of the expression product correlates with the presence or risk of prostate cancer, wherein the antigen-presenting cell is optionally formulated with a pharmaceutically acceptable carrier.